

Current Issues in Public Health Genomics



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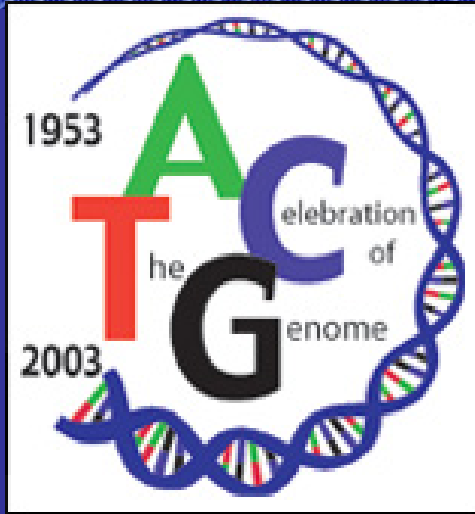


Overview

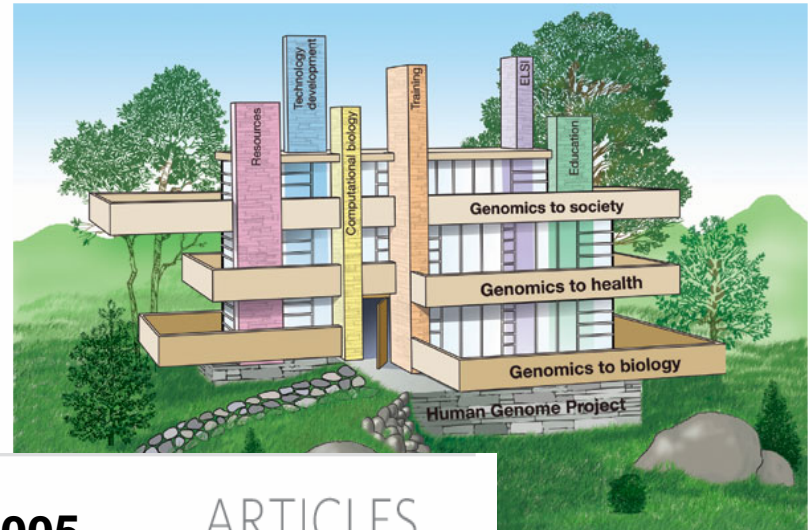
- Public Health in the Genomics Era
- Examples of CDC Public Health Genomics Building Block Initiatives
- Near Term Directions



Welcome to the Genomics Era!



URL of this page: <http://www.genome.gov/11007576>



Nature 2005

ARTICLES

A haplotype map of the human genome

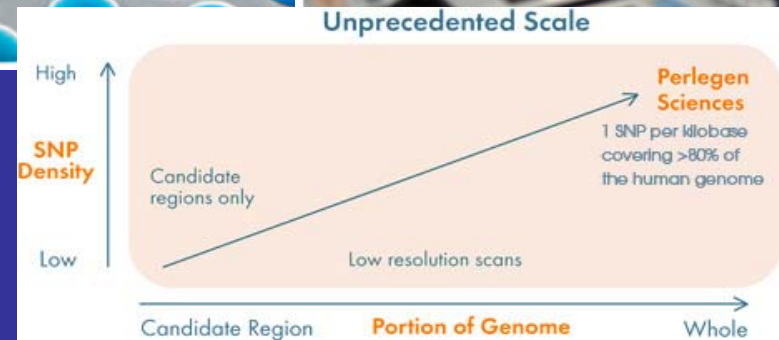
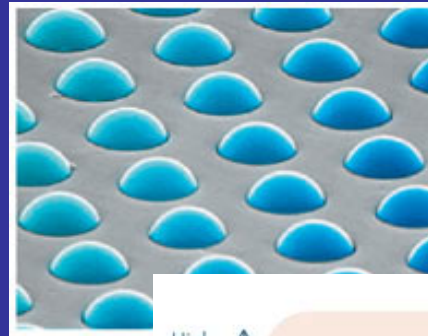
The International HapMap Consortium*

Inherited genetic variation has a critical but as yet largely uncharacterized role in human disease. Here we report a public database of common variation in the human genome: more than one million single nucleotide polymorphisms (SNPs) for which accurate and complete genotypes have been obtained in 269 DNA samples from four populations, including ten 500-kilobase regions in which essentially all information about common DNA variation has been extracted. These data document the generality of recombination hotspots, a block-like structure of linkage disequilibrium and low haplotype diversity, leading to substantial correlations of SNPs with many of their neighbours. We show how the HapMap resource can guide the design and analysis of genetic association studies, shed light on structural variation and recombination, and identify loci that may have been subject to natural selection during human evolution.

“The SNPs are down: genotyping for the rest of us”

- “Human genotyping has never been hotter, and a sophisticated set of array-based tools now simplifies the process dramatically, facilitating everything from small basic research studies to complex genetic epidemiology.”

A. Dove. Nat Methods 2005;2, 989 - 994



Address http://www.fnih.org/GAIN/GAIN_home.shtml Go Link

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GENETIC ASSOCIATION INFORMATION NETWORK (GAIN)

The Genetic Association Information Network (GAIN) is a public-private partnership of the Foundation for the National Institutes of Health, Inc. (FNIH), which will include corporations, private foundations, advocacy groups, concerned individuals, and the National Institutes of Health (NIH) ([Overview](#)). This initiative will take the next step in the search to understand the genetic factors influencing risk for complex diseases. Through a series of whole genome association studies, using samples from existing case-control studies of common diseases, the project will contribute to the identification of genetic pathways that make us more susceptible to these diseases and thereby facilitate discovery of new molecular targets for prevention, diagnosis, and treatment.



“Gene Increases Diabetes Risk, Scientists Find”

(New York Times, January 16, 2005)

- “Scientists have discovered a variant gene that leads to a sizeable extra risk of Type 2 Diabetes and is carried by more than a third of Americans...An immediate practical consequence of the discovery would be to develop a diagnostic test to identify people who carry the variant gene. If people knew of their extra risk, they would have an incentive to stay thin and exercise.”

Grant SFA et al. Nat Genet Jan 2006

Address <http://www.diabetes.co.uk/news/2006/Jan/important-genetic-discovery-could-treat-diabetics.html>

Diabetes.co.uk
The Diabetes Portal

You are eligible to register if you are aged between 21 & 70 years of age and have diagnosed Type 2 diabetes mellitus

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Important genetic discovery could treat diabetics

Tue, 17 Jan 2006

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Perlegen Sciences
Leader in pharmacogenomics research

http://medicine.plosjournals.org/archive/1549-1676/3/2/pdf/10.1371_journal.pmed.0030114-S.pdf

94%

DOI: 10.1371/journal.pmed.0030109

Does Genetic Testing Really Improve the Prediction of Future Type 2 Diabetes?

A. Cecile J. W. Janssens, Marta Gwinn, Subramony Subramonia-Iyer, Muin J. Khoury

From their study on the genetic prediction of future type 2 diabetes (T2D), Lyssenko and colleagues conclude that "genetic testing might become a future approach to identify individuals at risk of developing T2D" [1]. One of their most striking findings is an impressive 21.2-fold increased risk for T2D in obese carriers of the *PPARG* PP and *CAPN10* SNP43/44 GG/TT genotypes with elevated fasting plasma glucose (FPG).

A closer look at their results reveals that the hazard ratio of 21.2 was obtained by comparing the T2D risks of persons who have all three risk factors ("risk genotypes," obesity, and elevated FPG) with those who have none of these factors. This hazard ratio, thus, measures the combined increase in risk due to *PPARG* PP and *CAPN10* SNP43/44 GG/TT genotypes, obesity, elevated FPG, and their interactions. Among obese persons with elevated FPG, the incidence of T2D was 44.7% in carriers of risk genotypes and 10.7% in persons with other genotypes, yielding a risk ratio of 4.2 (95% confidence interval [CI], 2.3–7.8; follow-up time, age, and sex were not taken into account), a result that is statistically significant.



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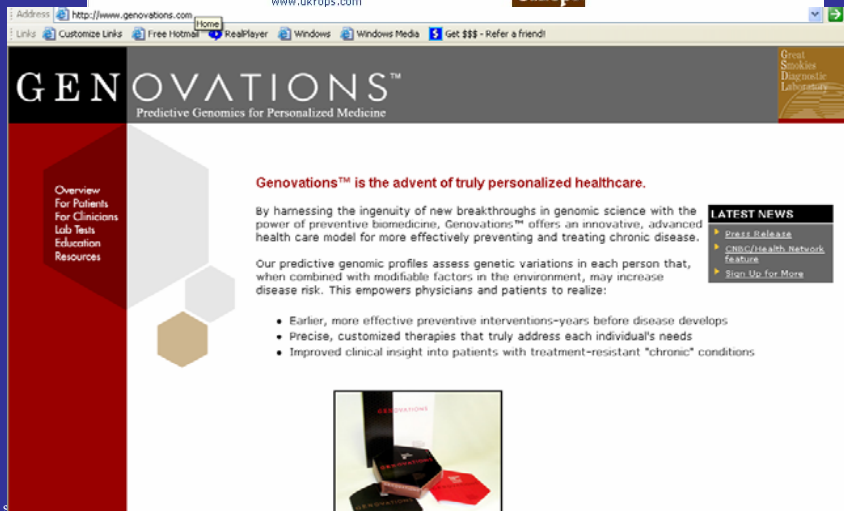
Ukrops

PRODUCTS

Roche Makes Waves with AmpliChip Launch

09/11/03—Reinforcing its position as a pharmacogenomics pioneer, diagnostic giant Roche launched in June a P450 chip measuring DNA markers for predicting patient responses to many common drugs.

The AmpliChip CYP450 test is based on the Affymetrix GeneChip DNA analysis platform. The chip detects variations in DNA that are known to affect genes controlling the body's mechanisms for processing drugs, and it is the first chip using Affymetrix technology that meets federal standards for clinical use. The test can be run only in reference laboratories, which must meet specific certification standards. In the future, though, it could become easier to use in a variety of settings.

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By harnessing the ingenuity of new breakthroughs in genomic science with the power of preventive biomedicine, Genovations™ offers an innovative, advanced health care model for more effectively preventing and treating chronic disease.

Our predictive genomic profiles assess genetic variations in each person that, when combined with modifiable factors in the environment, may increase disease risk. This empowers physicians and patients to realize:

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Office of In Vitro Diagnostic Device Evaluation and Safety (OIVD)



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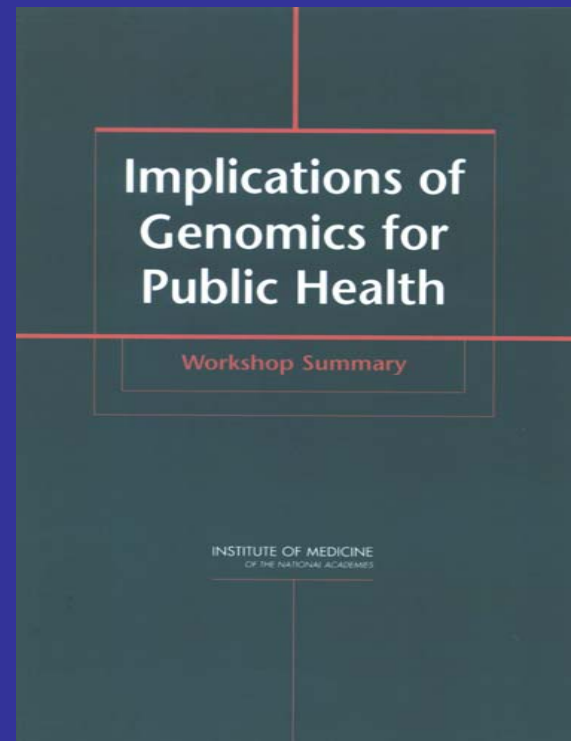
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OIVD Requests a Meeting with Roche Diagnostics Regarding the AmpliChip CYP450 Microarray

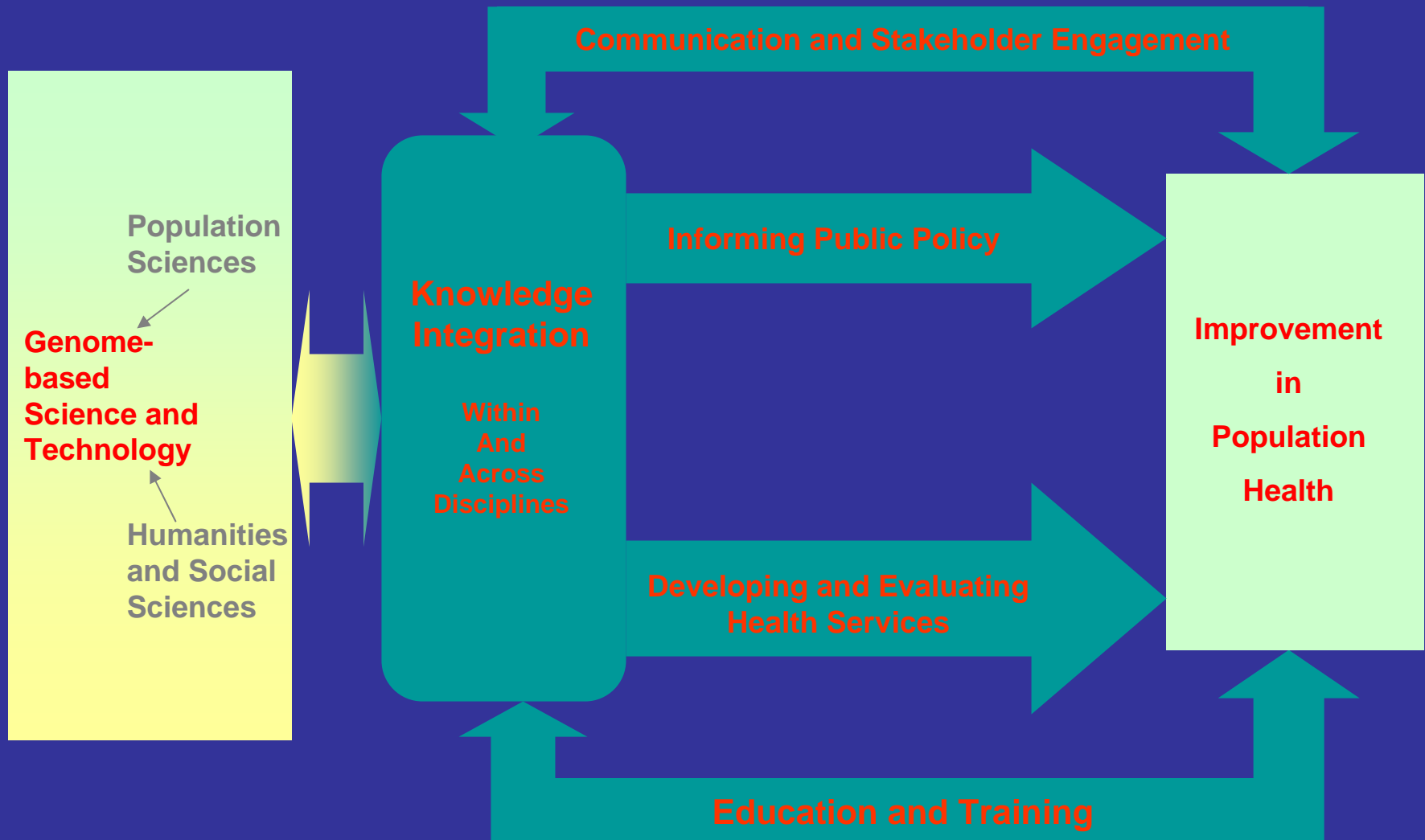
What is “Public Health Genomics”?

(Institute of Medicine, 2005)

- “An emerging field that assesses the impact of genes and their interaction with behavior, diet and the environment on the population’s health”



The Public Health Genomics Enterprise



Public Health Genomics at CDC

Examples of Building Block Initiatives

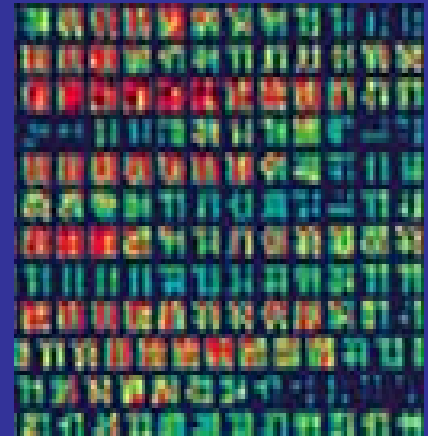
- Evaluation of Genomic Applications in Practice and Prevention (EGAPP)
- Family History Public Health Initiative
- National Profile of Genetic Variation
- Integrating Genomics into Public Health Investigations and Preparedness
- Public Health Genomics Response Capacity



Evaluation of Genomic Applications in Practice and Prevention (EGAPP)

Benefit: Build the Evidence Base

- Independent panel to evaluate genetic tests and other genomic applications
- Provide evidence base for practitioners and protect public from harm
- CDC accepts key leadership role recommended by many groups
- Extends CDC efforts in newborn screening and genetic testing quality assurance



Family History Public Health Initiative

Benefit: Tools for Prevention

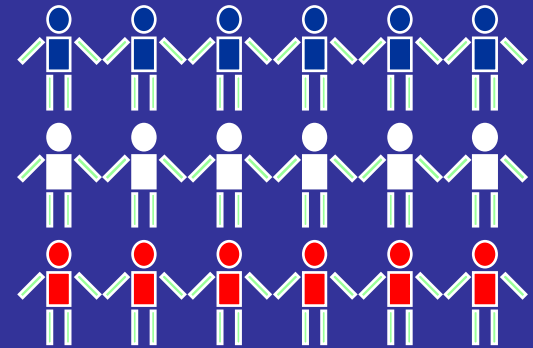
- Family history captures shared genes, behaviors, and environment
- Use to target screening, prevention
- CDC tool (6 diseases) & validation study in collaboration with CDC, NIH, academia
- Partnership with Surgeon General



National Profile of Genetic Variation

Benefit: Population Data for Health Impact

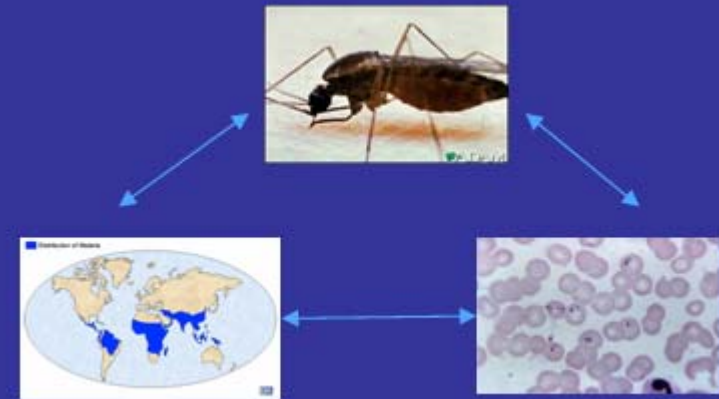
- NHANES: Representative sample of U.S. population with rich clinical, laboratory and exposure data
- Needed for estimating numbers of people at risk and who could benefit from interventions
- CDC working group projects:
 - Phase I: 100-200 variants
 - Phase II: 300,000-500,000 variants



Genomics in Public Health Investigations

Benefit: Prevent and Control Disease in Communities

- Pathogen genomics a key public health tool
- Human genomics: susceptibility, vaccine and drug response, adverse effects
- Identify environmental factors for intervention



Public Health Genomics Response Capacity

Benefit: Prepare public health capacity & workforce

- Centers for excellence in genomics & public health
- Integrating genomics into state programs
- Public health genomics workforce competencies
- Reports, website, online courses, weekly update

Want to Learn More about Genomics?

Six Weeks to Genomic Awareness is a free, online series of six modules designed to provide public health professionals a foundation for understanding how genomics advances are relevant to public health.

The series includes:

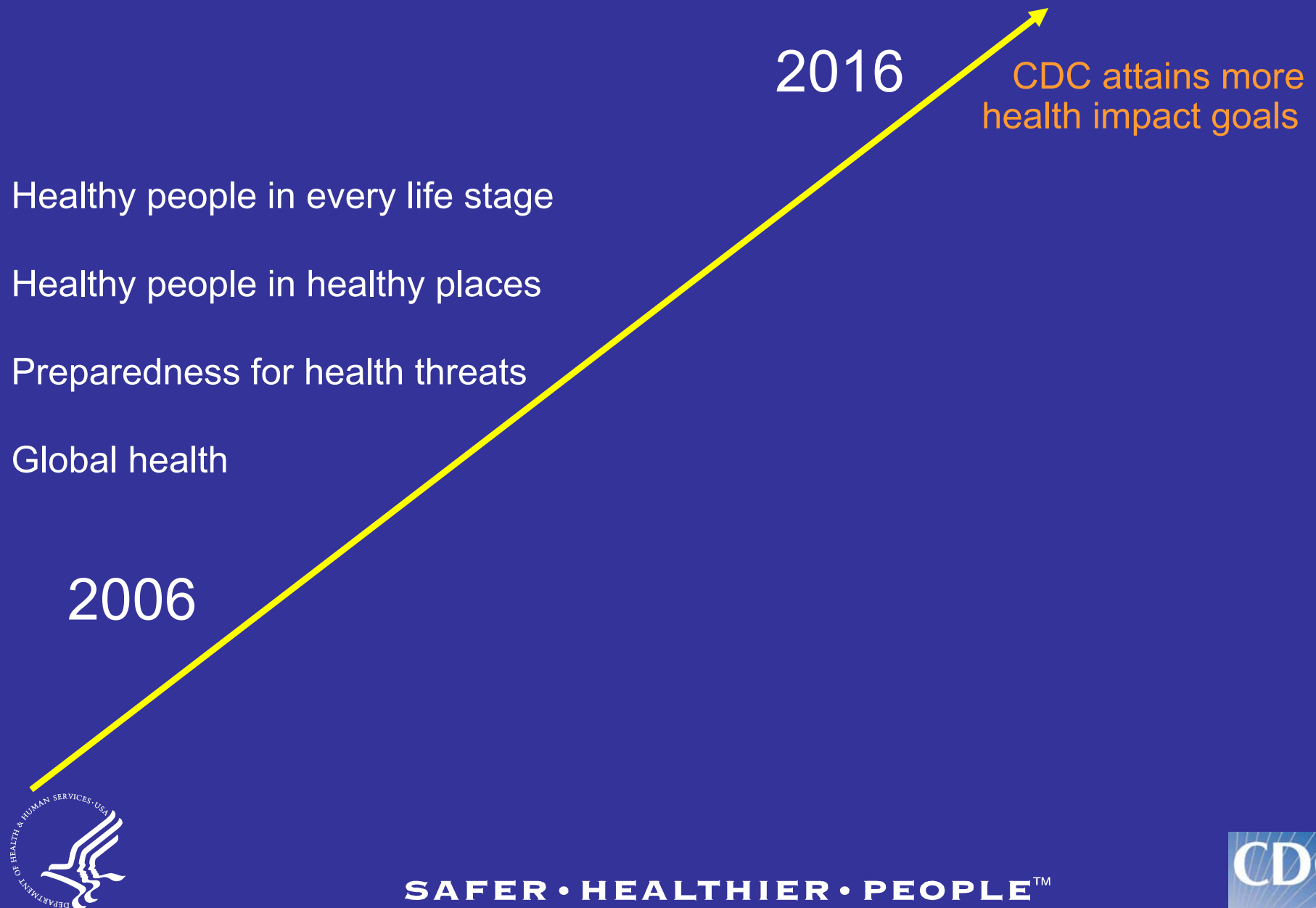
- Introductory Genomics Concepts
- Examination of Genes in Populations
- Genetic Testing
- Gene-Environment Interactions
- Ethical, Legal and Social Issues
- State and National Resources



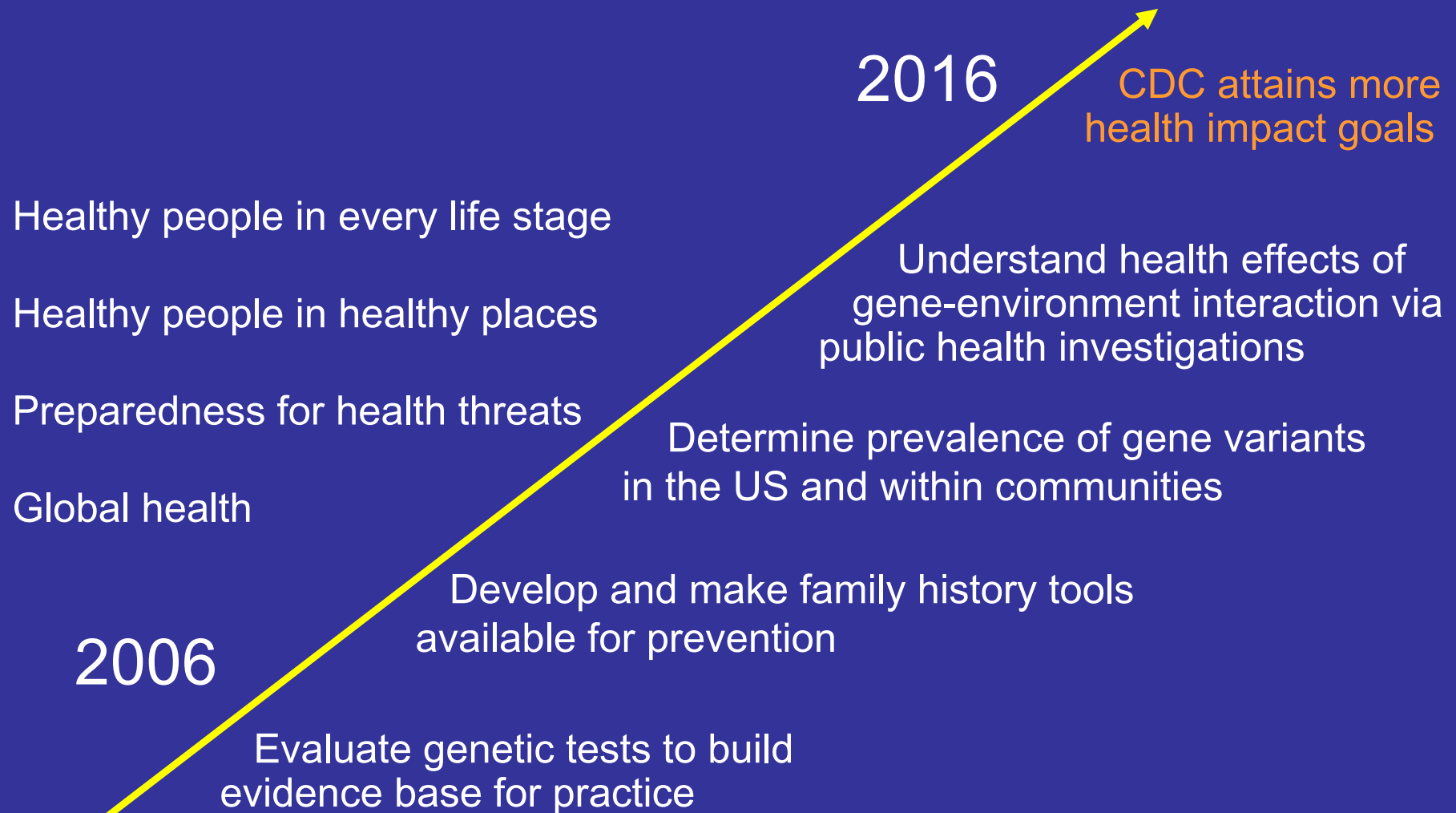
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Public Health Genomics: a Vision for the Future



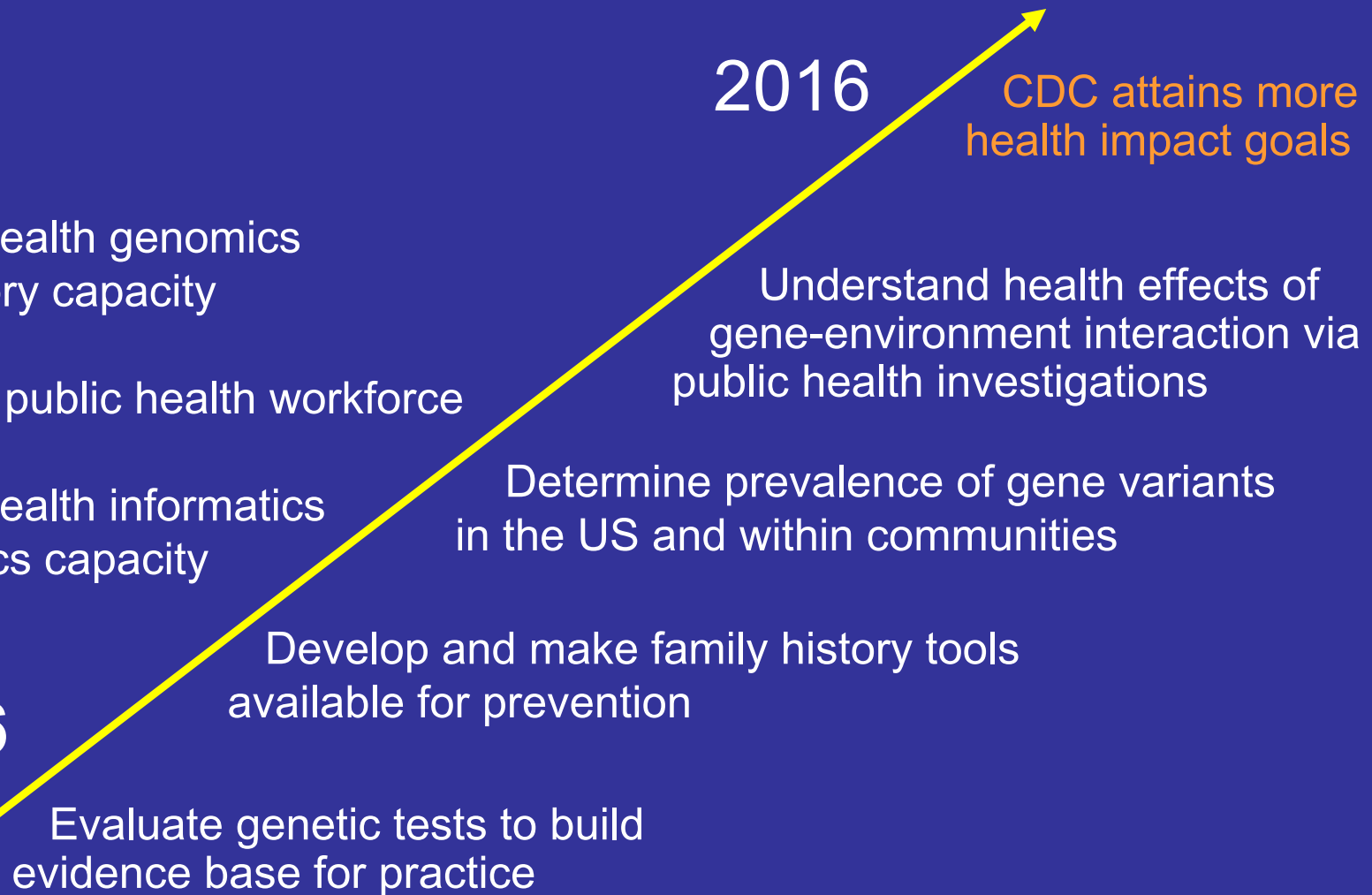
Public Health Genomics: a Vision for the Future



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Public Health Genomics: a Vision for the Future



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Near Term Directions in Public Health Genomics at CDC

- Enhancing networking and collaborations
- Developing CDC goals oriented initiatives
- Making seed funding available
- Emphasizing career development



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